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CASE REPORT

Chromosome 10q Deletion del (10)(q26.1q26.3) is Associated with Cataract

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Distal 10q deletion syndrome is an uncommon chromosomal disorder. Interstitial deletion involving bands 10q25—10q26.1 is extremely rare and few cases have been reported. The characteristic features are facial dysmorphisms, postnatal growth retardation, developmental delay, congenital heart disease, genitourinary anomalies, digital anomalies, and strabismus. We report for the first time a patient with *de novo* 10q interstitial deletion del (10)(q26.1q26.3) and cataract.

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1. Introduction

Patients with terminal deletion of the long arm of chromosome 10 present with phenotypic manifestations, including facial dysmorphisms, postnatal growth retardation, developmental delay, mental retardation, digital anomalies, cardiac defects, and genitourinary defects.[1,2] It is an uncommon chromosomal disorder, with most terminal deletions starting at breakpoints in bands 10q25 or 10q26. In contrast to these terminal deletions, interstitial deletions within bands 10q25—10q26.3 are extremely rare and only seven cases have been reported.[3] However, it is unknown whether the phenotypes are different from terminal deletions. Here, we report the first reported case with a *de novo* 10q interstitial deletion, del (10)(q26.1q26.3). In addition to many of the phenotypic anomalies previously described in interstitial 10q cases, our patient presented with cataracts.

It is estimated that there are 1.5 million blind children in the world.[4] Cataract is the main cause of treatable blindness in children. Information on the ocular and systemic characteristics of pediatric cataract syndromes is useful for further systemic screening needs and genetic evaluation.

2. Case Report

The patient, a 10.5-year-old girl, is the first child of healthy, unrelated parents. She was born spontaneously at 36 weeks with uncomplicated pregnancy. Her birth body weight was 2.25 kg (25th percentile). She had a triangular and asymmetric face, prominent nasal bridge, hypertelorism, malformed ears, right ear skin tag, simian crease, and bilateral clinodactyly (Figure 1A and B). Congenital heart disease with ventricular septal defect, patent ductus arteriosus, and severe pulmonary artery hypertension was found soon after birth. Total surgical repair was performed at 7 months. Evoked-response audiometry revealed a left sensorineural hearing impairment (hearing loss: left: 65 dB). Development has shown a global delay. She first sat at 14 months, crawled at 20 months, stood at 24 months, and began walking alone at 29 months of age. Her first meaningful words were spoken at 5 years of age and speech delay has been noted since, limited to a few words. Furthermore, verbal comprehension is limited to common verbal instructions. Growth delay was also noted at 6 months old, her height was 54.8 cm ($<3^{rd}$ percentile) and her weight was 3.8 kg ($<3^{rd}$ percentile). Global development assessment at 8 years and 10 months showed a developmental quotient (DQ) of 36. Cranio-cerebral magnetic resonance imaging (MRI) was normal. Renal sonography revealed bilateral kidney hypotrophy with no other urogenital defects. The patient is now 10 years and 5 months old. Her height is 103.5 cm ($<3^{rd}$ percentile) and her weight is 14.5 kg ($<3^{rd}$ percentile).

Eye examination revealed a cataract on the left side at 7 years of age, which was the reason for the first visit to our hospital. The left retina was normal and there was no strabismus.

Chromosome preparation of peripheral white blood cells was performed, and trypsin-banding Giemsa was applied at 550 band resolution. G-banding analysis revealed that the patient had a karyotype of 46, xx, del (10) (q26.1q26.3) (Figure 2). In order to rule out familial translocation, parental blood samples were also analyzed and found to be normal. These results indicated that the chromosomal aberration was *de novo* in origin. An additional study with fluorescence *in situ* hybridization (FISH) was performed (Figure 3). A single whole-chromosome painting probe specific for chromosome 10 (CPMC-GM10926) was employed

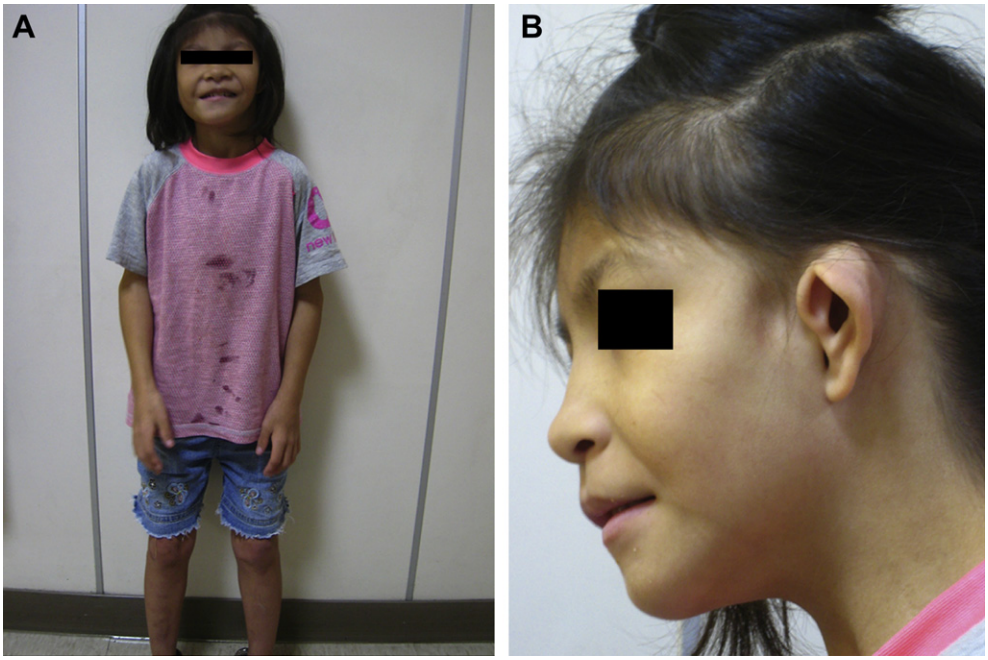

Figure 1 Gross appearance and face of this case. Note the triangular and asymmetric face, prominent nasal bridge, hypertelorism and malformed ear.